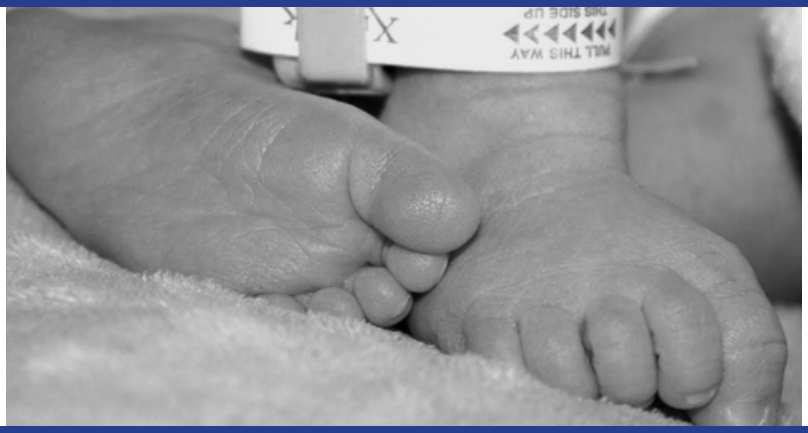
SCID, Angels for Life Foundation



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How Newborn Screening for Severe Combined Immune Deficiency will save the lives of Florida's youngest citizens

Florida has recently joined other states including Wisconsin, Massachusetts, California, New York, Puerto Rico, Colorado, Connecticut, Michigan, Delaware, Mississippi, Texas and Minnesota (January 2013) in screening all newborn infants for SCID (Severe Combined Immune Deficiency), a rare but fatal disorder of the immune system. On April 17, 2012 the Florida Governor authorized a Department of Health budget of nearly two-million dollars to begin this statewide program aimed at early detection and the treatment of SCID.

Virtually all babies born in the United States are tested for an array of treatable, but potentially deadly, conditions within 48 hours of birth, SCID is one of those treatable conditions. SCID was the 30th condition added to the existing recommended uniform screening panel. The genetic disorders are included in the test of dried blood spots obtained on filter paper from the heel stick of infants that is taken at birth. Infants with SCID, commonly known as "The Bubble Boy disease," generally die before the age of two years, if they are not treated. SCID is one of the few genetic conditions that can be cured with hematopoietic stem cell (or bone marrow) transplantation or, in some cases, even gene therapy. However, treatment must be initiated early, by 3.5 months of age, for it to be most effective. Another type of treatment that could be given is enzyme replacement therapy. However, this form of therapy is for infants who have the ADA-deficiency form of SCID.

What is SCID and why should Newborn Screening be implemented in the United States?

SCID is a primary immunodeficiency disease. Affected infants lack T and B lymphocytes, the white blood cells that help fight infections, and make up the adaptive immune system. Babies with SCID appear healthy at birth and most go unrecognized unless blood testing is ordered, specifically testing the immune system. Without testing, there is no way to tell if an infant has the condition until they begin to experience infections from viruses, bacteria, or fungi. Many cases go undetected until the children die of overwhelming opportunistic infections, without a diagnosis.

SCID babies can even develop disseminated infections from live vaccines such as the rotavirus vaccine, reinforcing the need for early diagnosis. As a result, SCID has been called a pediatric emergency. For more information on how the rotavirus vaccine can impact babies with SCID, please visit our website at www.SCIDAngelsforlife.com.

Research has shown that transplants performed prior to 3.5 months of life have an overall survival rate of 94 percent, but the survival rate drops to 69 percent for those transplanted after 3.5 months.

How are infants screened for SCID?

SCID screening in newborns became possible just a few years ago, with the development of a DNA based screening method. The test used for SCID screening detects whether or not the infant is producing enough of their own T cells by identifying TRECs (T-cell Receptor Excisison Circles). The test is inexpensive, about $17 per test in Florida, and that includes the necessary equipment, reporting and personnel to perform the screening on the assay. This is a highly sensitive and specific assay and, based on the results of states currently screening, the false positive rate is 0.018 percent and sensitivity is 99.8 percent. There are at least 17 different SCID genes and the vast majority of them will be detected using this newborn screening test.

Parents of infants who have abnormal screening, can go to our website at www.SCIDAngelsforlife.com and read "Newborn Screening for Severed Combined Immune Deficiency (SCID) and Conditions Associated with T Cell Lymphopenia" which was written for parents who receive a positive screening, this will explain the significance of the result and what next steps to expect. Infants whose screening shows fewer than 25 TREC copies per blood spot, will then need to have confirmatory

testing, using flow cytometry, to detect the presence of T cells, B cells, and NK cells in the blood. This is a standard, FDA approved, assay that is available in most hospital or reference laboratories. If SCID is confirmed, the infant will need to be started on antibiotics to prevent opportunistic infection and will be immediately referred to a center that has experience with SCID and can determine the best treatment option.

Receiving a positive diagnosis of SCID is a frightening event and we encourage families to go to our website at www.SCIDAngelsforlife.com and learn more about what it means to be diagnosed with SCID.

In Florida we are lucky that there are three treatment centers available to treat children diagnosed with SCID. They are: USF - All Children's Hospital in St. Petersburg, UM - University of Miami Hospital in Miami, and UF - Shand's Children's Hospital in Gainesville. However, All Children's Hospital is the only center in Florida actively transplanting children with SCID at this time (2012). The All Children's Hospital Outpatient Care Center (OCC) houses the University of South Florida's Division of Allergy, Immunology, and Rheumatology, where infants and children suspected of having SCID and other immune disorders are treated. The Sinus and Allergy Program at the University of Miami Miller School of Medicine is committed to providing comprehensive and state of the art patient care.

How SCID Angels has positively impacted the State

SCID, Angels For Life Foundation led the drive to have Newborn Screening for SCID implemented in our state. We spent countless hours lobbying and arranging multiple meetings with the Newborn Screening Advisory Committee, the Florida Surgeon General and the Governor's office, playing a key role in educating all of these departments about the importance of SCID. Sponsoring the legislation was Representative Matt Hudson, Florida District 101 in the Collier county area. Along with the guidance and expertise of Dr Elena Perez, University of Miami Miller School of Medicine, we were able to show the Department of Health and the Governor why newborn screening for SCID is cost effective and saves lives! Thank you to the above mentioned people for all their tireless efforts.

How common is SCID

The true incidence of SCID is not yet known, however, with more and more states implementing screening we are getting closer to learning what that number is. It has been reported in California that during their first six months of screening for SCID they found an incidence of 1 out of every 32,000 live births and 1 out of 22,000 in the Hispanic population. With approximately 220,000 live births in Florida per year, we can expect to see six to seven new cases each year.

FL detects its first case of SCID through newborn screening

Less than 3 weeks after beginning newborn screening for SCID in Florida an infant was identified by newborn screening and confirmed as having SCID. October 20, 2012 after approximately 19,000 babies in Florida had been screened, an innocent little girl was born with 0 TRECS. To sum up the process, in less than one week, a baby was born, screened for SCID, the family was notified of the positive screening, brought into the hospital lab for diagnostic evaluation, received confirmation of a positive case, and finally the family came in for their first consultation with the immunology and bone marrow transplantation teams. Only statewide newborn screening for SCID could make such a fast diagnosis a reality!